



NEWBORN SCREENING RESULT FOR BART'S HEMOGLOBIN

Physician's information sheet developed by the Nebraska Newborn Screening Program with review by James Harper, MD, Pediatric Hematologist with UNMC Follow-up program, and member of the Nebraska Newborn Screening Advisory Committee.

BACKGROUND

The alpha thalassemias result from the loss of alpha globin genes. There are normally four genes for alpha globin production so that the loss of one to four genes is possible.

The lack of one gene causes alpha thalassemia 2 (silent carrier) with no clinically detectable problems but may cause small amounts of hemoglobin Barts to be present in newborn blood samples. Alpha thalassemia trait (Alpha thalassemia 1) results from loss of two genes and causes a mild microcytic anemia which may resemble iron deficiency anemia. The loss of three genes causes hemoglobin H diseases which is a moderately severe form of thalassemia. The lack of all four genes causes hydrops fetalis and is usually fatal in utero.

In general, only the loss of one or two genes is seen in African Americans. Individuals from Southeast Asia and the Mediterranean may have all four types of alpha thalassemia.

The percentage of hemoglobin Barts in the blood sample may indicate the number of alpha genes that have been lost. However, the percentage of hemoglobin Barts is not directly measurable with the current methodology used by the newborn screening laboratory. Only the *presence* of Barts hemoglobin in relation to fetal and adult hemoglobin, and variants S, C, D and E can be detected.

RECOMMENDED WORK UP

In addition to the standard newborn hemoglobinopathy confirmation (hemoglobin electrophoresis), to separate those patients with alpha thalassemia silent carrier from the patients with alpha thalassemia trait, we recommend that these babies have the following labs drawn at their 6 month well baby check: CBC with retic count, ferritin, and a hemoglobin electrophoresis. The patient with alpha thalassemia silent carrier should have a normal CBC and retic, and a normal electrophoresis. A patient with alpha thalassemia trait will generally have a normal hemoglobin electrophoresis and retic, but microcytic red blood cells with an elevated red blood cell number. The hemoglobin in patients with alpha thalassemia trait may be slightly below normal to normal. The ferritin level reflects stored iron reserves. Many subtle hemoglobin defects like alpha thalassemia trait are not reliably diagnosed in the presence of iron deficiency. Documenting that the patient's iron level is normal improves the validity of the work up.

CLINICAL MANAGEMENT

Individuals with alpha thalassemia trait may have a very mild anemia with microcytosis and no other clinical problems. This anemia however, may be confused with iron deficiency anemia. Parents of infants with diagnosed hemoglobin Barts (alpha thalassemia trait), should be told it is inherited and that others in the family may have a similar disorder. They should be instructed to tell health professionals that their child has alpha thalassemia trait to avoid unnecessary tests or treatment with iron. These patients, like any child are capable of becoming iron deficient. Rather than relying on “trials of iron” or other empiric therapy, when iron deficiency is suspected in these children, it should be worked up and treated formally.

GENETIC COUNSELING

Family studies may be indicated after confirmatory testing of the newborn, to detect the more serious forms of alpha thalassemia. Particularly in those children who have a southeast Asian heritage, or in multiracial children, family studies may be useful as these ethnic groups have an increased risk for the more serious forms of alpha thalassemia and documenting this risk status may be useful in genetic counseling or family planning.

A basic patient information sheet is enclosed to aid in patient counseling. You may distribute to your patients as needed. If you have additional questions, please call Dr. James Harper of the pediatric hematology clinic at UNMC at 402-559-7257. Other hematology specialists are available at 402 955-3950 Dr. David Gnarra at Children's Hospital, or 308 762-2125 Dr. Howard Koch in Alliance.